A 15-year-old Nicaraguan female diagnosed with Proteus Syndrome at 3 months old presented to a pediatric cardiologist at 5 years of age after experiencing a near-syncopal event involving pallor, dizziness, and nausea. Her medical history was only significant for multiple surgical resections of soft tissue tumors in the umbilical and in the vaginal areas. Family history was negative for any congenital or acquired cardiac disease. On examination, swelling was noted of the right pectoral area, and there was right-sided hemihypertrophy. The remainder of the examination was normal, with a heart rate of 100 and blood pressure of 100/60 mm Hg. An initial electrocardiogram revealed a low atrial rhythm with right-axis deviation, right ventricular hypertrophy with strain, and ST changes in the inferior leads. An echocardiogram showed abnormal right ventricular thickening with abnormal diastolic filling. No valvular dysfunction or flow turbulence was noted. The patient was treated with β-blockers, restricted from competitive sports and physical education, and asymptomatic from a cardiorespiratory standpoint. At 6 years of age, she developed a seizure disorder, which was well controlled with medication and ultimately discontinued. Transthoracic (Figure 1) and transesophageal echocardiography (Figures 2 and 3) demonstrated thickening, marked echogenicity, and signal inhomogeneity of the biventricular walls, and no outflow tract obstruction was observed.

A cardiac MRI was performed at 15 years of age to evaluate the progressive ventricular wall thickening seen by serial echocardiography. There was no evidence of structural heart disease or left ventricular outflow tract obstruction. The MRI was remarkable for marked biventricular and septal hypertrophy with inhomogeneity of signal intensity. Figure 4A and 4B shows midventricular 4-chamber and short-axis steady state free precession cine images, gated to diastole. In both images, there is hyperintense/bright signal of the tissue surrounding and between the left and right ventricles when compared with the normal myocardial signal intensity at the endocardial surface. Figure 5A and 5B shows T1-weighted turbo spin echo; these slices also demonstrate a markedly hyperintense signal of the same epicardial and midseptal segments. After application of a fat saturation pulse, these same segments have been described, but there have been few reports describing cardiac disease associated with Proteus syndrome. The first such case report was of a 20-year-old Chilean man with macrodactyly, partially amputated right index finger, cranial exostoses, proptosis, severe pulmonary fibrocystic changes, sinus
tachycardia, and right bundle branch block. His echocardiogram demonstrated thickening of the cardiac septum and an apical right ventricular mass, but no tissue characterization or histology was obtained. The presence of fatty elements has been described by cardiac MRI in the cardiac tumor of a Japanese girl with multiple cranial hyperostoses and features suggestive of but not classic for Proteus Syndrome. This patient had a mass localized to the anterior right ventricular free wall with bright signal on T1-weighted images, similar to our patient.

The degree of cardiac lipomatosis, as we describe in this case, has never been reported in the literature. Cardiac MRI was critical in defining tissue characteristics but equally important in providing quantitative data that were able to define normal cardiac volumes, mass, and function, separate from the lipomatous tissue. Certainly, the clinical significance of this diagnosis is unclear because some pediatric cardiac tumors, including lipomatous masses, have been associated with ventricular arrhythmias and aborted sudden death and may result in tumor erosion through the myocardium. Most recently, this patient underwent defibrillator placement after a few episodes of syncope with no documented arrhythmias by Holter monitoring. However, an electrophysiology study was positive for inducible ventricular tachycardia.

Disclosures
None.

References

Keywords: cardiomyopathy, hypertrophic ▼ hypertrophy ▼ lipomatosis ▼ magnetic resonance imaging ▼ pediatric
Figure 5. Four-chamber (A) and short-axis (B) sequences gated to end-diastole. A and B, Turbo spin echo T1-weighted images, which reveal fatty elements as hyperintense signal surrounding normal myocardium. The right coronary artery can be seen coursing through the lipomatous tissue (**). Note that the thin arrow in A demonstrates the serous pericardium.

Figure 6. Four-chamber (A) and short-axis (B) sequences gated to end-diastole. A and B, Turbo spin echo images with application of a fat saturation pulse; the fatty tissue is suppressed and thus hypointense. Note that the thin arrow in A demonstrates the serous pericardium.

Figure 7. Four-chamber (A) and short-axis (B) sequences gated to end-diastole. A and B, Diffuse and heterogeneous gadolinium uptake in the lipomatous tissue consistent with a fibrotic component to the mass. Again note that there is septal involvement (s) best seen not only on these late gadolinium enhancement images but also throughout the 4 sequences. Note that the thin arrow in A demonstrates the serous pericardium.

Figure 8. A cine short-axis stack of the ventricles obtained in end-diastole. Endocardial contours of the left ventricle (yellow), right ventricle (red), and epicardial contours (blue) are used to calculate end-diastolic volumes and myocardial mass for each ventricle. Similar contours were performed for the ventricular endocardial borders in systole to calculate stroke volume and ejection fractions for both ventricles, as listed in the Table. A fourth contour, in green, surrounds the lipomatous tissue. The ventricular volumes and muscle volumes were subtracted from the lipoma contour volume to obtain a volume of fat. This value was multiplied by the specific gravity of fat (0.9) to obtain an estimate of lipoma mass, which was 494 g/m² when compared with a combined ventricular mass of 80.3 g/m².

Table. MRI-Derived Ventricular Volumes, Function, and Mass

<table>
<thead>
<tr>
<th></th>
<th>LV</th>
<th>RV</th>
<th>Lipomatous Tissue</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>EF, %</strong></td>
<td>65.1</td>
<td>66.9</td>
<td>...</td>
</tr>
<tr>
<td><strong>EDVi, mL/m²</strong></td>
<td>86.2</td>
<td>82</td>
<td>...</td>
</tr>
<tr>
<td><strong>ESVi, mL/m²</strong></td>
<td>30.1</td>
<td>27.1</td>
<td>...</td>
</tr>
<tr>
<td><strong>SVi, mL/m²</strong></td>
<td>56.1</td>
<td>54.9</td>
<td>...</td>
</tr>
<tr>
<td><strong>Mass, g/m²</strong></td>
<td>51.4</td>
<td>28.9</td>
<td>494</td>
</tr>
<tr>
<td><strong>Cardiac index</strong></td>
<td>3.98</td>
<td>...</td>
<td>...</td>
</tr>
</tbody>
</table>

EDVi indicates end-diastolic volume indexed; EF, ejection fraction; ESVi, end-systolic volume indexed; LV, left ventricle; Massi, mass indexed; RV, right ventricle; and SVi, stroke volume indexed.
Are You Calling Me Fat? An Extreme Case of Cardiac Lipomatosis Masquerading as Hypertrophic Cardiomyopathy
Eva H. Nunlist, Jorge Garcia and Ruchira Garg

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SUPPLEMENTAL MATERIAL

Video 1 is a transesophageal echocardiogram (TEE) clip of the left ventricle showing marked hypertrophy and echogenicity of the myocardium, but no significant left ventricular outflow tract obstruction.

Video 2 is a TEE clip of the right ventricle showing involvement of the right ventricular free wall and conal septum as well.

Video 3 is a steady state free precession (SSFP) cine MRI image in the 4 chamber plane. In this movie, the difference in signal from the normal myocardium (hypointense/dark) and the lipomatous tissue (hyperintense/bright) is readily apparent.

Video 4 is a SSFP cine MRI image in short axis at the mid-ventricular level showing marked concentric biventricular lipomatous tissue, including infiltration of the mid-septum. Biventricular systolic function is clearly preserved.